

**CLAIMS**

1. A method for determining whether a subject has, or is at risk of developing, an abnormally low HDL level, comprising determining the identity of the allelic variant of a polymorphic region of the SR-BI gene of the subject and comparing the allelic variant of the subject with allelic variants associated with abnormally low HDL levels, to thereby determine whether the subject has an allelic variant of a polymorphic region of an SR-BI gene associated with a abnormally low HDL level.
2. A method of claim 1, wherein the polymorphic region is located in an exon.
3. A method of claim 2, wherein the intron is exon 8.
4. A method of claim 3, wherein the polymorphic region is a nucleotide polymorphism.
5. A method of claim 4, wherein the nucleotide polymorphism is located at position 41 of exon 8.
6. A method of claim 5, wherein nucleotide 41 of exon 8 of the SR-BI gene in a normal subject is a thymidine and the presence of a nucleotide other than a thymidine at position 41 of exon 8 in the SR-BI gene of a subject indicates that the subject has or is at risk of developing an abnormally low HDL level.
7. A method of claim 6, wherein the nucleotide other than a thymidine at position 41 of exon 8 is a cytidine.
8. A method of claim 1, wherein determining the identity of the allelic variant of a polymorphic region of an SR-BI gene comprises determining the identity of at least one nucleotide of the polymorphic region.

9. A method of claim 2, wherein determining the identity of the allelic variant of a polymorphic region comprises contacting a nucleic acid of the subject with at least one probe or primer which is capable of hybridizing to an SR-BI gene.
- 5 10. A method of claim 9, wherein the probe or primer is capable of specifically hybridizing to an allelic variant of the polymorphic region.
- 10 11. A method of claim 10, wherein the probe or primer is capable of specifically hybridizing to an allelic variant having a thymidine at position 41 of exon 8 of the SR-BI gene.
12. A method of claim 1, wherein the probe or primer has a nucleotide sequence from about 15 to about 30 nucleotides.
- 15 13. A method of claim 1, wherein the probe or primer is a single stranded nucleic acid.
14. A method of claim 1, wherein the probe or primer is labeled.
- 20 15. A method of claim 1, wherein determining the identity of the allelic variant of a polymorphic region is carried out by allele specific hybridization.
16. A method of claim 1, wherein determining the identity of the allelic variant of a polymorphic region is carried out by primer specific extension.
- 25 17. A method of claim 1, wherein determining the identity of the allelic variant of a polymorphic region is carried out by an oligonucleotide ligation assay.
18. A method of claim 1, wherein determining the identity of the allelic variant of a polymorphic region comprises performing a restriction enzyme site analysis.
- 30

19. A method of claim 18, wherein the restriction enzyme is a HaeIII enzyme.
20. A method of claim 1, wherein determining the identity of the allelic variant of a polymorphic region is carried out by single-stranded conformation polymorphism.
- 5 21. A method for determining whether a female subject has, or is at risk of developing, an abnormally low HDL level, comprising determining the identity of the allelic variant of a polymorphic region of the SR-BI gene of the subject and comparing the allelic variant of the subject with allelic variants associated with low HDL levels, to
- 10 thereby determine whether the subject has or is at risk of developing an abnormally low HDL level.
22. A method of claim 21, comprising determining the identity of the nucleotide at 41 in exon 8 and/or nucleotide 54 in intron 5, wherein the presence of a cytidine at position
- 15 41 of exon 8 and/or the presence of a thymidine at position 54 of intron 5 indicates that the subject has or is at risk of developing an abnormally low HDL level.
23. A kit for determining whether a subject has, or is at risk of developing, a low HDL level, comprising a probe or primer which is capable of hybridizing to an SR-BI gene and thereby identifying whether the SR-BI gene contains an allelic variant of a
- 20 polymorphic region which is associated with a low HDL level and instructions for use in diagnosing a subject as having, or having a predisposition, towards developing a low HDL level.
- 25 24. A kit of claim 23, wherein the polymorphic region is located in an exon.
25. A kit of claim 24, wherein the exon is exon 8.
26. A kit of claim 25, wherein the polymorphic region is a nucleotide polymorphism
- 30 located at nucleotide 41 of exon 8.

27. A kit of claim 26, wherein the presence of a cytidine at nucleotide 41 of exon 8 of the SR-BI gene is indicative that the subject has or is at risk of developing an abnormally low HDL level.

5 28. A kit of claim 23, wherein the polymorphic region is located in an intron, and wherein the subject is female.

29. A kit of claim 28, wherein the intron is intron 5.

10 30. A kit of claim 29, wherein the polymorphic region is a nucleotide polymorphism located at nucleotide 54 of intron 5.

15 31. A kit of claim 30, wherein the presence of a thymidine at nucleotide 54 of intron 5 of the SR-BI gene is indicative that the subject has or is at risk of developing an abnormally low HDL level.

20 32. A kit for determining whether a female subject has, or is at risk of developing, an abnormally low HDL level, comprising a probe or primer which is capable of hybridizing to an SR-BI gene and thereby identifying whether the SR-BI gene contains an allelic variant of a polymorphic region which is associated with a low HDL level and instructions for use in diagnosing a subject as having, or having a predisposition, towards developing a low HDL level.

25 33. A kit of claim 32, wherein the polymorphic region is a nucleotide selected from the group consisting of nucleotide 41 in exon 8 and nucleotide 54 in intron 5, wherein the presence of, the presence of a cytidine at position 41 of exon 8 and/or the presence of a thymidine at position 54 of intron 5 indicates that the subject has or is at risk of developing an abnormally low HDL level.

30 34. A method for predicting the effect of hormone replacement therapy on the HDL level in a female subject comprising: identifying one or more allelic variants of the SR-BI

gene which are associated with abnormally low HDL levels in females, thereby predicting the effect of hormone replacement therapy on the HDL level in the subject.

5 35. The method of claim 34, wherein hormone replacement therapy results in an abnormally low HDL level.

36. The method of claim 34, wherein the allelic variants comprise a cytidine at position 41 of exon 8 and/or a thymidine at position 54 of intron 5.

10 37. The method of claim 34, wherein the female subject is postmenopausal.

15 38. A method of predicting the effect of hormone replacement therapy on a female subject, wherein the identification of allelic variants of the SR-B1 gene which are associated with abnormally low HDL levels in females results in a prediction that hormone replacement therapy will result in abnormally low HDL levels.

COPIES OF THIS DOCUMENT ARE ON FILE IN THE RECORDS SECTION OF THE PATENT AND TRADEMARK OFFICE